INVENTOR(S): JOHN P. CARULLI ET AL.

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FIG. 1A FIG. 1B

FIG. 1

ATTORNEY'S DOCKET NO: 032796-217 High Bone Mass Trait Clear Symbol Symbol definitions Unaffected -L@034-40.000 -4004-0 38 35 94, Q 

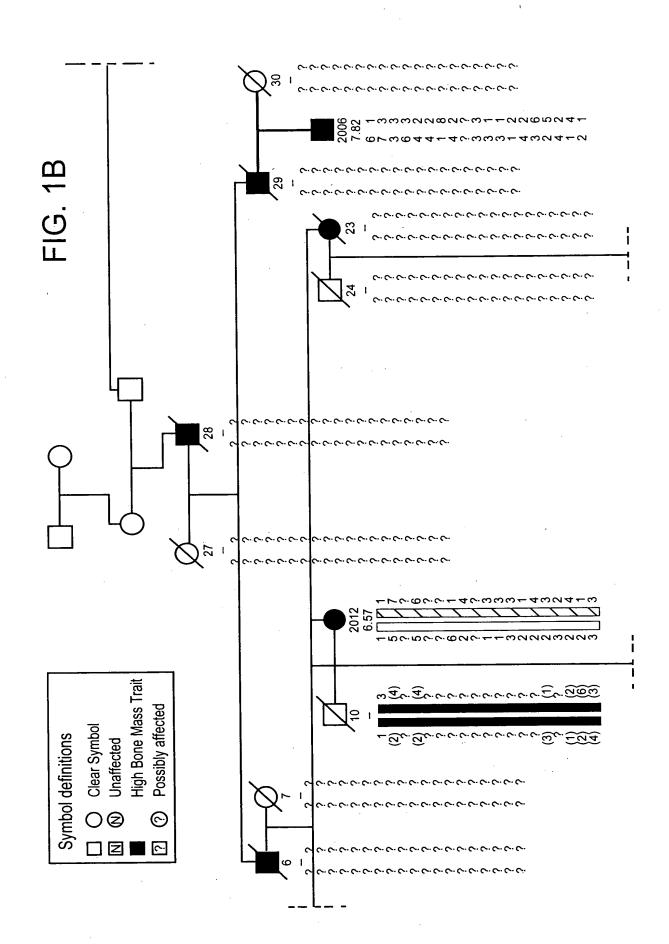
APPLN. FILING DATE: DECEMBER 10, 2003 TITLE: HIGH BONE MASS GENE OF 11Q13.3

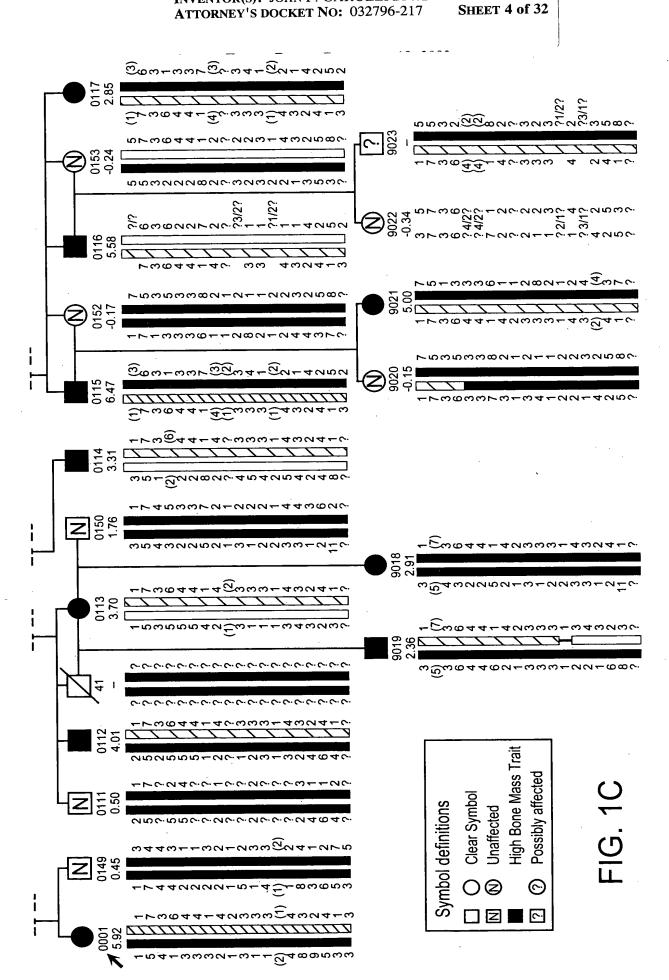
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**INVENTOR(S):** JOHN P. CARULLI ET AL.

INVENTOR(S): JOHN P. CARULLI ET AL.

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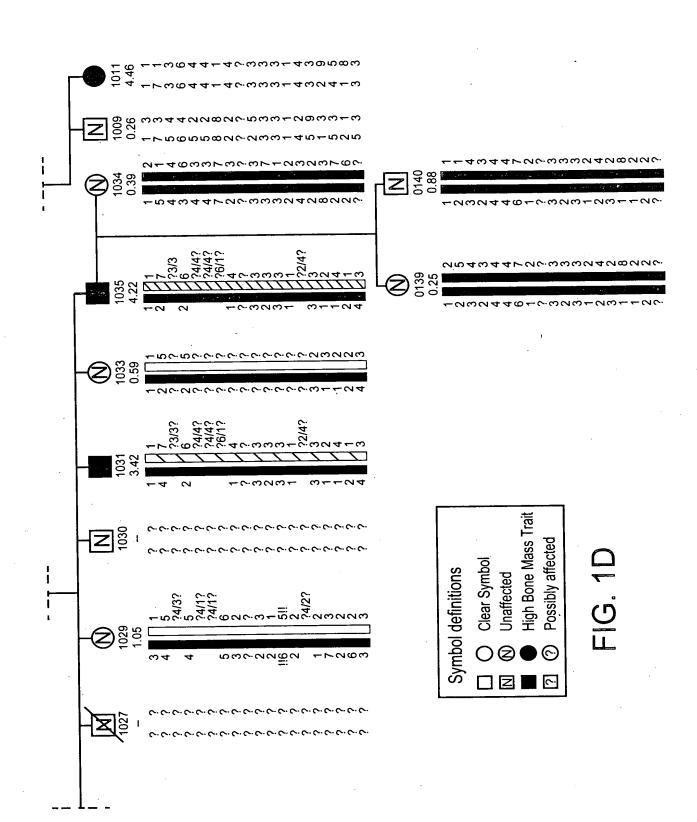


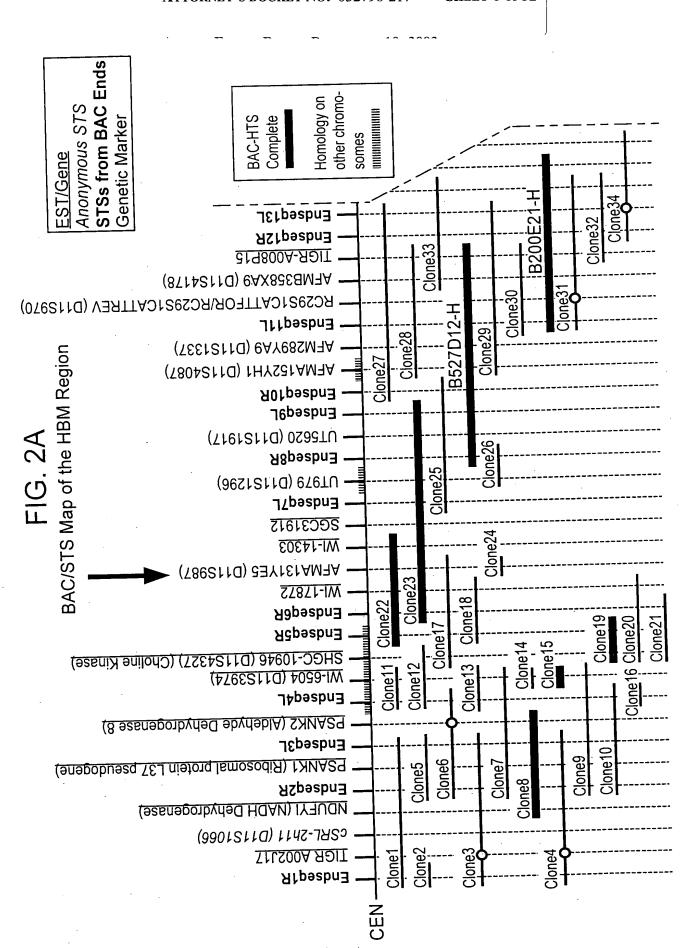


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SHEET 7 of 32 ATTORNEY'S DOCKET NO: 032796-217 2HCC-11930 FGF3PCR1 (Fibroblast Growth Factor 3) YFMB032ZG5 (D11S4136) Clone 79 (1884311a) 6916-IM Clone74 Clone 77 FGF4 (Fibroblast Growth Factor 4) Clone75 Clone78 CI11-424 (D112008) CCND1 (Cyclin D1) Endseq32L Clone 76 Clone63 D112468 Clone62 Clone72 P117F3T Clone 73 Clone61 BCF(1)/BCF1(S) (B Cell F\u00e4mbpoma) Clone60 %Endseq31R BAC/STS Map of the HBM Region Clone7 &Endseq30R (GS) **Я6Zpasbn∃**% Clone56 Clone 70 Endseq28R (GS) Clone58 STSs from BAC Ends **SEndseq27**L FIG. 2B Endseq26R (GS) Anonymous STS Genetic Marker ⊗Endseq25∟ (GS) SEndseq24R Clone50 EST/Gene **Endseq23R** Clone69 Clone51 Clone 55 lone53 Clone67 Endseq22L (GS) %Endseq21R **20031049** (76211G) '2-132Mq Clone49 CC111-44A Endseq20L Clone48 Clone68 Endseq19R Clone66 **ATrpssbn3** Clone46 Clone45 AFMA272YB5 (D11S4113) Clone44 2HCC-4167 (NIBS43) (SMBP2) Clone43 Clone65 **Endseq16R** Clone39 Clone40 Clone,41 Clone42 MI-2875 (D11S4404) Clone38 Clone37 Clone 36 WI-11974 (Carnitine Palroiteyl Transferase) GALN (Preprogalanin) Clone35 Endseq15L Endseq14R other chromo-Homology on (363612110) 2821/1821 BAC-HTS Complete somes

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Exon 1 CCGTGGCCGCTGCTGCTGCTGCTGCTGCTGCCGCC TGCCCGGCCCCCGCGGCC

Exon 2 Coordinates: 527d12\_Contig308G 30944-30549 gcccacagCCTCGCCGCTCCTGCTATTTGCCAACCGCCGGGACGTACGGC TGGTGGACGCCGGCGGAGTCAAGCTGGAGTCCACCATCGTGGTCAGC GGCCTGGAGGATGCGCCGCAGTGGACTTCCAGTTTTCCAAGGGAGC GAACCAGACGGGGGCCGCCGTGCAGAACGTGGTCATCTCCGGCCTGG TCTCTCCCGACGCCTCGCCTGCGACTGGGTGGGCAAGAAGCTGTACT GGACGGACTCAGAGACCAACCGCATCGAGGTGGCCAACCTCAATGGC ACATCCCGGAAGGTGCTCTTCTGGCAGGACCTTGACCAGCCGAGGGCC ATCGCCTTGGACCCCGCTCACGGgtaaaccctgctg

... 9408 nt ...

Exon 3 Coordinates: 527d12\_Contig308G 21141-20945

 ${\tt ccccgtcacagGTACATGTACTGGACAGACTGGG} \underline{\underline{G}} {\tt TGAGACGCCCCGGATTG}$ AGCGGGCAGGGATGGATGGCAGCACCCGGAAGATCATTGTGGACTCG GACATTTACTGGCCCAATGGACTGACCATCGACCTGGAGGAGCAGAAG CTCTACTGGGCTGACGCCAAGCTCAGCTTCATCCACCGTGCCAACCTG **GACGGCTCGTTCCG**gtaggtacccac

... 6094 nt ...

Exon 4 Coordinates: 527d12\_Contig308G 15047-14850 tccctgactgcag GCAGAAGGTGGTGGAGGCCAGCCTTCGCCCTGACGCTCTCCGGGGACACTCTGTACTGGACAGACTGGCAGACCCGC TCCATCCATGCCTGCAACAAGCGCACTGGGGGGAAGAGGAAGAT CCTGAGTGCCCTATACTCACCCATGGACATCCAGGTGCTGAGCCAGGA **GCGGCAGCCTTTCT**gtgagtgccgg

... 1827 nt ...

Exon 5 Coordinates: 527d12\_Contig308G 13220-13088 tttctcagTCCACACTCGCTGTGAGGAGGACAATGGCGGCTGCTCCCACCTG TGCCTGCTGCCCCAAGCGAGCCTTTCTACACATGCGCCTGCCCCACG GGTGTGCAGCTGCAGGACAACGGCAGGACGTGTAAGGCAGgtgaggcggtgg gacg

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... 20923 nt ...

Exon 6 Coordinates: 527d12\_Contig309G 7705-8100
ctccacagGAGCCGAGGAGGTGCTGCTGCTGGCCCGGCGGACGGACCTAC
GGAGGATCTCGCTGGACACGCCGGACTTCACCGACATCGTGCTGCAGG
TGGACGACATCCGGCACGCCATTGCCATCGACTACGACCCGCTAGAGG
GCTATGTCTACTGGACAGATGACGAGGTGCGGGCCATCCGCAGGGCG
TACCTGGACGGGTCTGGGGCGCAGACGCTGGTCAACACCGAGATCAA
CGACCCCGATGGCATCGCGGTCGACTGGGTGGCCCGAAACCTCTACTG
GACCGACACGGGCACGGACCGCATCGAGGTGACGCGCCTCAACGGCA
CCTCCCGCAAGATCCTGGTGTCGGAGGACCTGGACGACCCCGAGCC
ATCGCACTGCACCCCGTGATGGGgttaagacgggc

..... 3211 nt .....

..... 13445 nt .....

Exon 8 Coordinates: 527d12\_Contig309G 24927-25143
ccgtcctgcagGTGATCAATGTTGATGGGACGAAGAGGCGGACCCTCCTGGA
GGACAAGCTCCCGCACATTTTCGGGTTCACGCTGCTGGGGGACTTCAT
CTACTGGACTGACTGGCAGCGCCGCAGCATCGAGCGGGTGCACAAGG
TCAAGGCCAGCCGGGACGTCATCATTGACCAGCTGCCCGACCTGATGG
GGCTCAAAGCTGTGAATGTGGCCAAGGTCGTCGgtgagtccggggggtc

....2826 nt .....

Exon 9 Coordinates: 527d12\_Contig309G 27969-28256
gttcgcttccagGAACCAACCCGTGTGCGGACAGGAACGGGGGGGTGCAGCCA
CCTGTGCTTCTTCACACCCCACGCAACCCGGTGTGGCTGCCCCATCGG
CCTGGAGCTGCTGAGTGACATGAAGACCTGCATCGTGCCTGAGGCCTT
CTTGGTCTTCACCAGCAGAGCCGCCATCCACAGGATCTCCCTCGAGAC
CAATAACAACGACGTGGCCATCCCGCTCACGGGCGTCAAGGAGGCCTC
AGCCCTGGACTTTGATGTGTCCAACAACCACATCTACTGGACAGACGT
CAGCCTGAAGgtagcgtgggc

.....3102.....

INVENTOR(S): JOHN P. CARULLI ET AL. ATTORNEY'S DOCKET NO: 032796-217 **SHEET 10 of 32** Exon 10 Coordinates: 527d12\_Contig309G 31358-31582 cctgctgccagACCATCAGCCGCGCCTTCATGAACGGGAGCTCGGTGGAGCA CGTGGTGGAGTTTGGCCTTGACTACCCCGAGGGCATGGCCGTTGACTG GATGGGCAAGAACCTCTACTGGGCCGACACTGGGACCAACAGAATCGA AGTGGCGCGGCTGGACGGCAGTTCCGGCAAGTCCTCGTGTGGAGGG ACTTGGACAACCCGAGGTCGCTGGCCCTGGATCCCACCAAGGGgtaagtgtt tgcctgtc .....1297 nt..... Exon 11 Coordinates: 527d12\_Contig309G 32879-33064 gtgccttccagCTACATCTACTGGACCGAGTGGGGCGGCAAGCCGAGGATCG TGCGGGCCTTCATGGACGGGACCAACTGCATGACGCTGGTGGACAAG GTGGGCCGGCCAACGACCTCACCATTGACTACGCTGACCAGCGCCTC TACTGGACCGACCTGGACACCAACATGATCGAGTCGTCCAACATGCTG Ggtgagggccgggct ..... 2069 nt..... Exon 12 Coordinates: 527d12\_Contig309G 35133-35454 gtgttcatgcagGTCAGGAGCGGGTCGTGATTGCCGACGATCTCCCGCACCCG TTCGGTCTGACGCAGTACAGCGATTATATCTACTGGACAGACTGGAAT CTGCACAGCATTGAGCGGCCGACAAGACTAGCGGCCGGAACCGCAC CCTCATCCAGGGCCACCTGGACTTCGTGATGGACATCCTGGTGTTCCA CTCCTCCCGCCAGGATGGCCTCAATGACTGTATGCACAACAACGGGCA GTGTGGGCAGCTGTGCCTTGCCATCCCCGGCGCCCACCGCTGCGGCT GCGCCTCACACTACACCCTGGACCCCAGCAGCCGCAACTGCAGCCgtaag tgcctcatggt ......2006 nt..... Exon 13 Coordinates: 527d12\_Contig309G 37460-37659

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.....6965 nt.....

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Exon 14 Coordinates: 527d12\_Contig309G 44624-44832 ctttgtcttacagCCCTTTGTTTTGACCTCTCTGAGCCAAGGCCAAAACCCAGAC AGGCAGCCCCACGACCTCAGCATCGACATCTACAGCCGGACACTGTTC TGGACGTGCGAGGCCACCAATACCATCAACGTCCACAGGCTGAGCGG GGAAGCCATGGGGGTGCTGCTGCGTGGGGACCCGCGACAAGCCCAGGG CCATCGTCGTCAACGCGGAGCGAGCGAGGGTAGGGGAGCGAGGGTAGGGGAGCGAGGGTAGGGGAGCGAGGGTAGGGGAGCGAGGGTAGGGGAGCGAGGGTAGGGGAGCGAGGGTAGGGGAGCGAGGGAGCGAGGGTAGGAGCCAAGCCCAGGG

.....1404 nt.....

Exon 15 Coordinates: 527d12\_Contig309G 46236-46427
ccaccetcccgcagGTACCTGTACTTCACCAACATGCAGGACCGGGCAGCCAA
GATCGAACGCGCAGCCCTGGACGCACCGAGCGCGAGGTCCTCTTCA
CCACCGGCCTCATCCGCCCTGTGGCCCTGGTGGTGGACAACACACTGG
GCAAGCTGTTCTGGGTGGACGCGGACCTGAAGCGCATTGAGAGCTGT
GACCTGTCAGgtacgcgccccgg

.....686 nt.....

Exon 16 Coordinates: 527d12\_Contig309G 47113-47322
ggctgcttgcagGGCCAACCGCCTGACCCTGGAGGACGCCAACATCGTGCA
GCCTCTGGGCCTGACCATCCTTGGCAAGCATCTCTACTGGATCGACCG
CCAGCAGCAGATGATCGAGCGTGTGGAGAAGACCACCGGGGACAAGC
GGACTCGCATCCAGGGCCGTGTCGCCCACCTCACTGGCATCCATGCAG
TGGAGGAAGTCAGCCTGGAGGAGTTCTgtacgtgggggc

.....3884 nt......

Exon 17 Coordinates: 527d12\_Contig309G 51206-51331 ttgtctttgcagCAGCCCACCCATGTGCCCGTGACAATGGTGGCTGCTCCCACA TCTGTATTGCCAAGGGTGATGGGACACCACGGTGCTCATGCCCAGTCC ACCTCGTGCTCCTGCAGAACCTGCTGACCTGTGGAGgtaggtgtgacctaggtgc

....3905 nt......

.....3052 nt.....

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Exon 19 Coordinates: 527d12_Contig309G 58524-58634 tctccttgcagCCATCTGCCTGCCCAACCAGTTCCGGTGTGCGAGCGGCCAGT GTGTCCTCATCAAACAGCAGTGCGACTCCTTCCCCGACTGTATCGACG GCTCCGACGAGCTCATGTGTGTGgtgagccagctt							
1448 nt							
Exon 20 Coordinates: 527d12_Contig309G 60082-60319 gtttgtctctggcagAAATCACCAAGCCGCCCTCAGACGACAGCCCGGCCCACA GCAGTGCCATCGGGCCCGTCATTGGCATCATCCTCTCTCT							
1095 nt							
Exon 21 Coordinates: 527d12_Contig309G 61414-61552 cttccctgccagGCATCGCATGCGGAAAGTCCATGATGAGCTCCGTGAGCCTGATGGGGGGGCCGGGGGGGG							
6513 nt							
Exon 22 Coordinates: 527d12_Contig309G 68065-68162 ttggctctcctcagATCCTGAACCCGCCGCCCTCCCCGGCCACGGACCCCTCCC TGTACAACATGGACATGTTCTACTCTTCAAACATTCCGGCCACTGCGA GACCGTACAGgtaggacatcccctgcag							

FIG. 3E

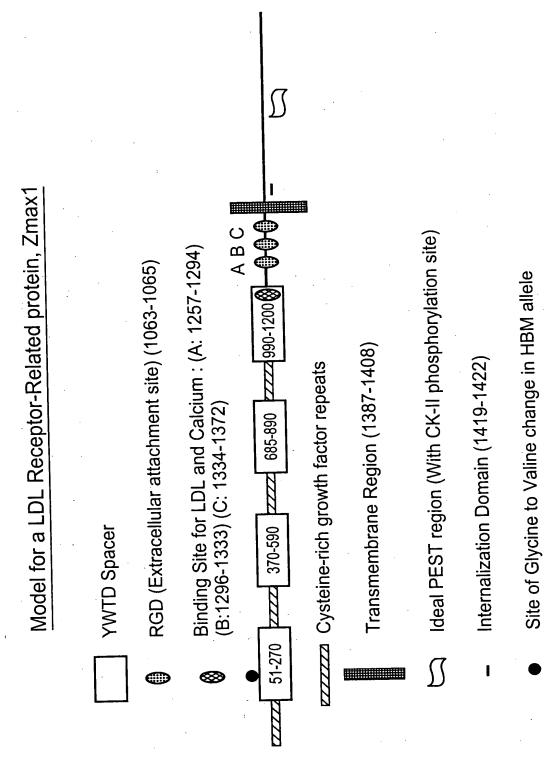
......2273 nt.....

INVENTOR(S): JOHN P. CARULLI ET AL.

FIG. 3F

INVENTOR(S): JOHN P. CARULLI ET AL.

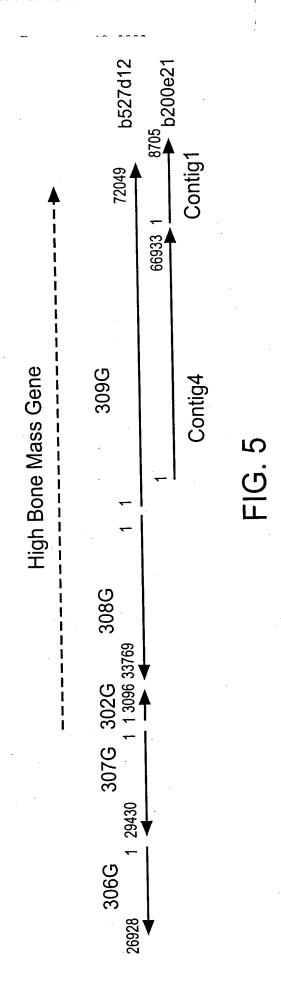
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C	S	)
(	F	)
	L	

60 120 17	180 37	240	300	360	420	480	540 157	600
ACTAAAGCGCCGCCGCCCATGGAGCCCGAGTGAGCGCGGCGCGGGCCCGTCCGGCC GCCGGACAACATGGAGGCAGCGCCGCCGGGCCGCCGTGGCGGCCGCTGCTGCTGCTGCT	GCTGCTGCTGCGGCTGCCCGGCCCCCGCGGCCTCGCCGCTCCTGCTATT L L L A L C G C P A P A A S P L L L F	TGCCAACCGCGGGACGTACGGCTGGTGGACGCCGGCGGAGTCAAGCTGGAGTCCACCAT A N R R D V R L V D A G G V K L E S T I	CGTGGTCAGCGGCCTGGAGGATGCGCCGCAGTGGACTTTTCCAAGGGAGCCGT	GTACTGGACAGACGTGAGGAGGCCATCAAGCAGACCTACCT	CGCCGTGCAGAACGTGGTCTCTCCCCGACGGCCTCGCCTGCGACTG A V Q N V V I S G L V S P D G L A C D W	$\mathtt{GGTGGGCAAGAAGCTGGACGGACTCAGAGACCGAACCGCATCGAGGTGGCCAACCT}$ V G K K L Y W T D S E T N R I E V A N L	CAATGGCACATCCCGGAAGGTGCTCTTCTGGCAGGACCTTGACCAGCCGAGGGCCATCGC N G T S R K V L F W Q D L D Q P R A I A	CTTGGACCCCGCTCACGGGTACATGTACTGGACAGACTGGGGTGAGACGCCCCGGATTGA L D P A H G Y M Y W T D W G E T P R I E
1 6 1	121 18	181 38	241 58	301 78	361 98	421 118	481 138	541 158

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197	217	r 780 237	2 840 257	2 900	297	C 1020 317	A 1080 337	T 1140
GCGGGCAGGATGGCAGCACCCGGAAGATCATTGTGGACTCGGACATTTACTGGCC R A G M D G S T R K I I V D S D I Y W P	CAATGGACTGACCTGGAGGAGCAGAAGCTCTACTGGGCTGACGCCAAGCTCAG N G L T I D L E E Q K L Y W A D A K L S	CTTCATCCACCGTGCCACCTGGACGGCTCGTTCCGGCAGAGGTGGTGGAGGGCAGCCT F I H R A N L D G S F R Q K V V E G S L	GACGCACCCTTCGCCCTCTCCGGGGACACTCTGTACTGGACAGACTGGCAGAC T H P F A L T L S G D T L Y W T D W Q T	CCGCTCCATCCATGCCTGCAAGCGCACTGGGGGGAAGAAGGAGGAGATCCTGAGTGC	CCTCTACTCACCATGGACATCCAGGTGCTGAGCCAGGAGCGGCAGCCTTTCTTCCACACACA	TCGCTGTGAGGAC R C E E	L TITCTACACATGCGCCTGCCCACGGGTGTGCAGCTGCAGGACACGGCAGGACGTGTAA	L GGCAGGAGCCGAGGTGCTGCTGCTGGCCGGCGGACGTACGGAGGATCTCGCT
601	661 198	721 218	781 238	841 258	901 278	961 298	1021 318	1081 338

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1680 537	L CTACTGGGGAGACCAAGACAAGATCGAGGTGATCAATGTTGATGGGACGAAGAG 3 Y W G D A K T D K I E V I N V D G T K R	1621 518
1620	_	1561 498
1560		1501 478
1500	CCTGGACGAGCCCATCGCACTGCACCCGTGATGGGCCTCATGTACTGGACAGA	1441 458
1440	GGACCGCATCGAGGTGACGCCTCAACGCACCTCCCGCAAGATCCTGGTGTCGGAGGA D R I E V T R L N G T S R K I L V S E D	1381 438
1380		1321
1320	CCGCAGGGCGTACCTGGACGGGTCTGGGCGCGAGACGAGATCAACGA R R A Y L D G S G A Q T L V N T E I N D	1261 398
1260 397		1201 378
1200 377	GGACACGCCGGACTTCACCGACATCGTGCAGGTGGACGACATCCGGCACGCCATTGC D T P D F T D I V L Q V D D I R H A I A	1141

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1681 538	GCGGACCCTCCTGGAGGACAAGCTCCCGCACATTTTCGGGTTCACGCTGCTGGGGGACTT R T L L E D K L P H I F G F T L L G D F	1740 557
1741	CATCTACTGGACTGGCAGCGCCGCAGCATCGAGCGGGTGCACAGGTCAAGGCCAG I Y W T D W Q R R S I E R V H K V K A S	1800 577
1801 578	CCGGGACGTCATTGACCAGCTGCCCGACCTGATGGGGCTCAAAGCTGTGAATGTGGC R D V I I D Q L P D L M G L K A V N V A	1860 597
1861 598	CAAGGTCGTCGGAACCCGTGTGCGGACAGGAACGGGGGGTGCAGCCACCTGTGCTT $_{ m K}$ $_{ m V}$ $_{ m V}$ $_{ m G}$ $_{ m T}$ $_{ m N}$ $_{ m C}$ $_{ m R}$ $_{ m N}$ $_{ m G}$ $_{ m G}$ $_{ m C}$ $_{ m S}$ $_{ m H}$ $_{ m L}$ $_{ m C}$ $_{ m F}$	1920 617
1921 618	CTTCACACCCCACGCAACCCGGTGTGGCTGCCCCATCGGCCTGGAGCTGAGTGACAT F T P H A T R C G C P I G L E L L S D M	1980
1981 638	GAAGACCTGCATCGTGCCTTCTTGGTCTTCACCAGCAGCCGCCATCCACAG K T C I V P E A F L V F T S R A A I H R	2040
2041 658	GATCTCCCTCGAGACCAATAACAACGACGTGGCCATCCCGCTCACGGGCGTCAAGGAGGC ISLETNNNDVAIPLTGVKEA	2100
2101 678	CTCAGCCCTGGACTTTGATGTGTCCAACAACCACATCTACTGGACAGACGTCAGCCTGAA S A L D F D V S N N H I Y W T D V S L K	2160 697
2161 698	GACCATCAGCCGCCTTCATGAACGGGAGCTCGGTGGAGCTTGGCCTTTGGCCTTTTGGCCTTTTGGCCTTTTGGCCTTTTGGCCTTTTGGCCTTTTGGCCTTTTGGCCTT	2220 717

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2280 737	2340 757	2400	2460 797	2520 817	2580	2640 857	2700 877	2760 897
TGACTACCCCGAGGGCATGGCCGTTGACTGGATGGGCAAGAACCTCTACTGGGCCGACAC D Y P E G M A V D W M G K N L Y W A D T	TGGGACCAACAGAATCGAAGTGGCGGCGGCAGGTTCCGGCAAGTCCTCGTGTG G T N R I E V A R L D G Q F R Q V L V W	GAGGGACTTGGACACCCGAGGTCGCTGGCTCCCACCAAGGGCTACATCTACTG R D L D N P R S L A L D P T K G Y I Y W	GACCGAGTGGGGCGGCAAGCCGAGGATCGTGCGGGCCTTCATGGACGGGCCCAACTGCAT T E W G G K P R I V R A F M D G T N C M	GACGCTGGTGGACAAGGTGGGCCGGGCCAACGACCTCACCATTGACTACGCTGACCAGCG	CCTCTACTGGACCGCCAACATGATCGAGTCGTCCAACATGCTGGGTCAGGA	GCGGGTCGTGATTGCCGACGATCTCCCGCACCTCCGGTCTGACGCAGTACAGCGATTA R V V I A D D L P H P F G L T Q Y S D Y	TATCTACTGGACAGCATCTGCACAGCATTGAGCGGGCCGACAAGACTAGCGGCCG	
2221 718	2281 738	2341 758	2401 778	2461 798	2521 818	2581 838	2641 858	2701 878

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2820 917	2880 937	2940 957	3000	3060 997	3120	3180	3240	3300
CTCCCGCCAGGATGGCCTCAATGACTGTATGCACAACAACAACGGGCAGTGTGGGCAGCTGTG S R Q D G L N D C M H N N G Q C G Q L C	CCTTGCCATCCCGGCGGCCACCGCTGCGGCTGCGCCTCACACTACACCCTGGACCCCAG	CAGCCGCAACTGCCGCCCCACCACCTTCTTGCTGTTCAGCCAGAAATCTGCCATCAG S R N C S P P T T F L L F S Q K S A I S	TCGGATGATCCCGGACGACCAGCCCGGATCTCATCCTGCCCTGCATGGACTGAG	GAACGTCAAAGCCATCGACTATGACCCACTGGACAAGTTCATCTACTGGGTGGATGGGCG	. CCAGAACATCAAGCGAGCCAGCCCCCTTTGTTTGACCTCTCTGAG	L CCAAGGCCAAAACCCAGACAGCCACGACCTCAGCATCGACATCTACAGCCGGAC	1 ACTGITCIGGACGIGCGAGGCCACCAATACCATCAACGICCACAGGCTGAGCGGGGAAGC 8 L F W T C E A T N T I N V H R L S G E A	1 CATGGGGGTGCTGCGTGGGGACCGCGACAAGCCCAGGGCCATCGTCGTCAACGCGGA 3 M G V V L R G D R D K P R A I V V N A E
2761 898	2821 918	2881 938	2941 958	3001	3061 998	3121	3181 1038	3241 1058

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3360	3420 1117	3480 1137	3540	3600	3660 1197	3720	3780 1237	3840
GCGAGGGTACCTGTACTTCACCAACATGCAGGACCGGGCAGCCAAGATCGAACGCGCAGC R G Y L Y F T N M Q D R A A K I E R A A	CCTGGACGCCACGCGCGCCTCTTCACCACCGGCCTCATCCGCCCTGTGGCCCT $_{ m L}$ D G T E R E V L F T T G L I R P V A L	GGTGGTGGACACACACTGGCTGTTCTGGGTGGACGCGGACCTGAAGCGCATTGA $_{ m V}$ $_{ m V}$ $_{ m D}$ $_{ m A}$ $_{ m D}$ $_{ m R}$ $_{ m L}$ $_{ m E}$ $_{ m V}$ $_{ m V}$ $_{ m D}$ $_{ m A}$ $_{ m D}$ $_{ m L}$ $_{ m K}$ $_{ m L}$ $_{ m E}$	GAGCTGTGACCTGGGGCCAACCGCCTGACCTGGAGGACGCCAACATCGTGCAGCC $S$ C D L $S$ G A N R L T L E D A N I V Q P	TCTGGGCCTGACCATCCTTCTACTGGATCGACCGCCAGCAGCAGATGAT L G L T I L G K H L Y W I D R Q Q Q M I	CGAGCGTGTGGAGAGACCAGGGGACAAGCGGACTCGCATCCAGGGCCGTGTCGCCCA E R V E K T T G D K R T R I Q G R V A H	CCTCACTGGCATCCAGGGGGGGAGTCAGCCTGGAGGAGTTCTCAGCCCACCCA	TGCCCGTGACAATGGTGGCTGCTCCCACATCTGTATTGCCAAGGGTGATGGGACACCACG A R D N G G C S H I C I A K G D G T P R	GTGCTCATGCCCAGTCCACTCGTGCTGCTGCTGACCTGTGGAGAGCCGCC $_{ m C}$ $_{ $
3301 1078	3361	3421	3481 1138	3541 1158	3601	3661	3721 1218	3781 1238

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3900	3960	4020 1317	4080	4140	4200	4260	4320 1417	4380
CACCTGCTCCCCGGACCAGTTTGCATGTGCCACAGGGGAGATCGACTGTATCCCCGGGGC T C S P D Q F A C A T G E I D C I P G A	CTGGCGCTGTGACGGCTTTCCCGAGTGCGATGACCAGAGGGCGGGGCTGCCCCGT	GTGCTCCGCCGCCCAGTTCCCCTGCGCGCGCGGGTCAGTGTGTGGACCTGCGCCTGCGCTG	CGACGGCGAGGCTGTCAGGACCGCTCAGACGAGGTGGACTGTGACGCCATCTGCCT D G E A D C Q D R S D E V D C D A I C L	GCCCAACCAGTTCCGGTGTGCGGCCAGTGTGTCTCTCATCAAACAGCAGTGCGACTC P N Q F R C A S G Q C V L I K Q Q C D S	CTTCCCCGACTGTATCGACGCTCCGACGAGCTCATGTGTGAAATCACCAAGCCGCCCTC F P D C I D G S D E L M C E I T K P P S			GGCCAACGGGCCCTTCCCGCACGAGTATGTCAGCGGGACCCCGCACGTGCCCCTCAATTT A N G P F P H E Y V S G T P H V P L N F
3841 1258	3901	3961 1298	4021 1318	4081	4141 1358	4201 1378	4261 1398	4321 1418

INVENTOR(S): JOHN P. CARULLI ET AL. ATTORNEY'S DOCKET NO: 032796-217

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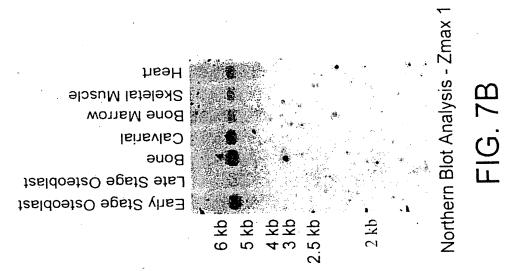
INVENTOR(S): JOHN P. CARULLI ET AL.

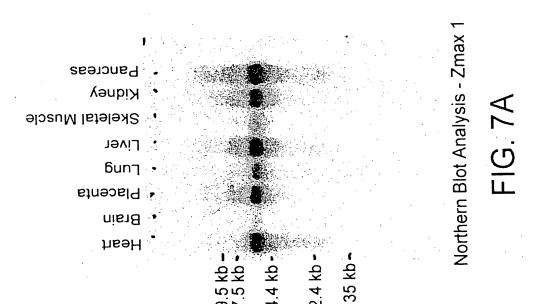
ATTORNEY'S DOCKET NO: 032796-217 SHEET 25 of 32

[3] [3]

INVENTOR(S): JOHN P. CARULLI ET AL.

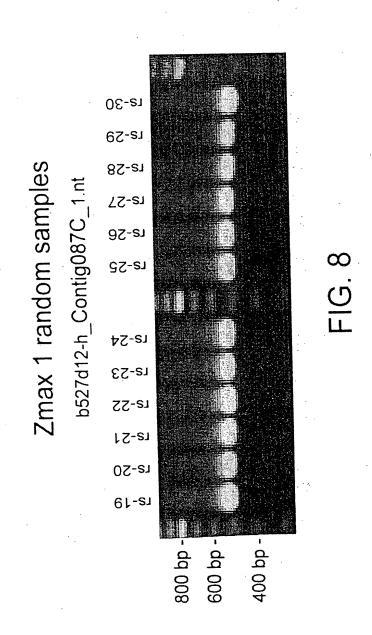
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INVENTOR(S): JOHN P. CARULLI ET AL.

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ASO Detection of the Zmax1 Exon 3 Mutation ග ග Affected A No pologia pologia Dology Brill <u>ග</u> ග **O O** Affected Affected A Kecked ᠐ᢆ᠐ b<sup>ejoejlen</sup>u  $\vdash 0$ Affected Delay ග ග Oligonucleotide Probes 2327 zmax1.ASO.t 2326 zmax1.ASO.g

INVENTOR(S): JOHN P. CARULLI ET AL.

ATTORNEY'S DOCKET NO: 032796-217 SHEET 29 of 32

# Mouse Zmax1 In situ hybridization 100X Magnification

### Antisense probe

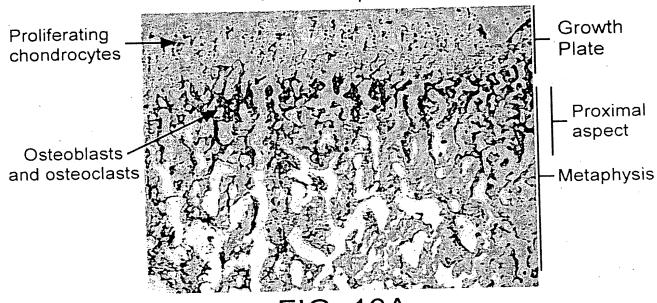


FIG. 10A

# Mouse Zmax1 In situ hybridization 100X Magnification

### Sense probe

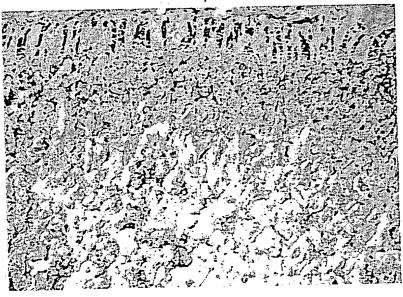


FIG. 10B

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ATTORNEY'S DOCKET NO: 032796-217 **SHEET 30 of 32** 

### Mouse Zmax1 In situ hybridization 400X Magnification Antisense probe

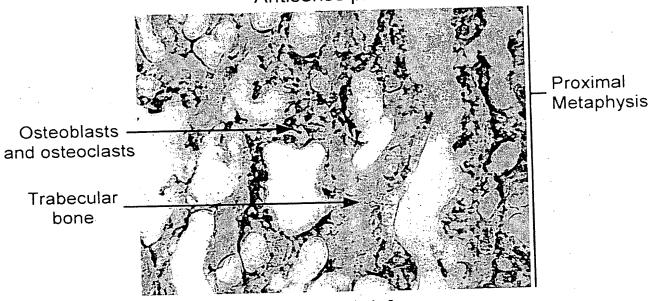


FIG. 11A

### Mouse Zmax1 In situ hybridization 400X Magnification Sense probe

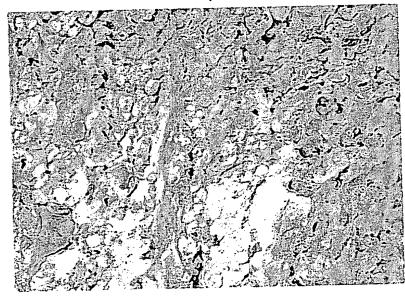
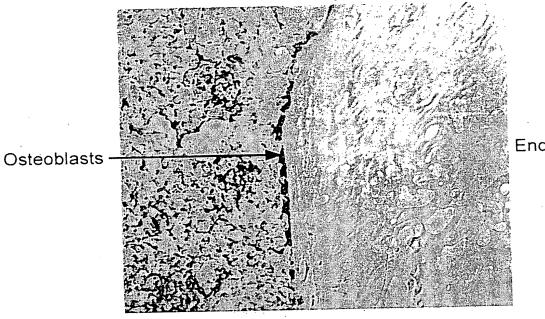


FIG. 11B

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### Mouse Zmax1 In situ hybridization 400X Magnification Antisense probe



Endosteum

FIG. 12A

### Mouse Zmax1 In situ hybridization 400X Magnification Sense probe

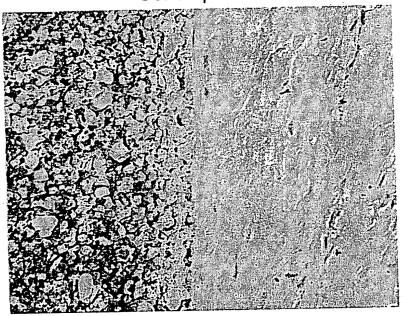
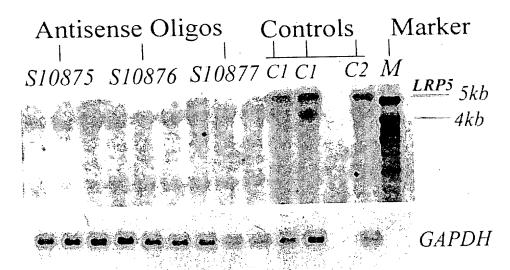


FIG. 12B

**INVENTOR(S):** JOHN P. CARULLI ET AL.

## Antisense Inhibition of Zmax1 Expression



MC-3T3 cells

FIG. 13